# Appendix 1. Non-pathogenic variants identified in patients from KwaZulu-Natal, South Africa, 2011-2021

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Variant | rs number | Effect | Pathogenicity | Number of patients |
| black | white | Indian  | coloured |
| NM\_007294.4(*BRCA1*):c.442-34C>T | rs799923 | Intron variant | Benign |  | 2 | 2 |  |
| NM\_007294.4(*BRCA1*):c.2597G>A, p.Arg866His | rs80356911 | Missense | Benign |  |  | 1 |  |
| NM\_007294.4(*BRCA1*):c.4113G>A, p.Gly1371= | [rs147448807](https://www.ncbi.nlm.nih.gov/snp/rs147448807) | Synonymous | Likely-benign |  |  |  | 1 |
| NM\_007294.4(*BRCA1*):c.4308T>C, p.Ser1436\_Ser1437= | [rs1060915](https://www.ncbi.nlm.nih.gov/variation/tools/1000genomes/?chr=17&from=41234470&to=41234470&gts=rs1060915&mk=41234470:41234470|rs1060915) | Synonymous | Benign |  |  | 6 | 1 |
| NM\_007294.4(*BRCA1*):c.4837A>G, p.Ser1613Gly | [rs1799966](https://www.ncbi.nlm.nih.gov/variation/tools/1000genomes/?chr=17&from=41223094&to=41223094&gts=rs1799966&mk=41223094:41223094|rs1799966) | Missense | Likely-benign |  |  | 6 | 1 |
| NM\_000059.4(*BRCA2*):c.-26G>A | [rs1799943](https://www.ncbi.nlm.nih.gov/snp/rs1799943) | 5' UTR | Benign |  | 1 | 1 |  |
| NM\_000059.4(*BRCA2*):c.425+67A>C | [rs11571610](https://www.ncbi.nlm.nih.gov/snp/rs11571610) | Intron variant | Benign |  | 1 | 2 |  |
| NM\_000059.4(*BRCA2*):c.426-89T>C | [rs3783265](https://www.ncbi.nlm.nih.gov/snp/rs3783265) | Intron variant | Benign |  | 1 | 1 |  |
| NM\_000059.4(*BRCA2*):c.517-4C>G | [rs81002804](https://www.ncbi.nlm.nih.gov/variation/tools/1000genomes/?chr=13&from=32900632&to=32900632&gts=rs81002804&mk=32900632:32900632|rs81002804) | Intron variant | Likely-benign |  |  |  | 2 |
| NM\_000059.4(*BRCA2*):c.681+56C>T | [rs2126042](https://www.ncbi.nlm.nih.gov/snp/rs2126042) | Intron variant | Benign |  | 1 | 3 | 1 |
| NM\_000059.4(*BRCA2*):c.865A>C, p.Asn289His | [rs766173](https://www.ncbi.nlm.nih.gov/snp/rs766173) | Missense | Benign |  | 1 | 2 |  |
| NM\_000059.4(*BRCA2*):c.1114A>C, p.Asn372His | [rs144848](https://www.ncbi.nlm.nih.gov/snp/rs144848) | Missense | Benign |  |  | 3 |  |
| NM\_000059.4(*BRCA2*):c.1365A>G, p.Ser455= | [rs1801439](https://www.ncbi.nlm.nih.gov/snp/rs1801439) | Synonymous | Benign |  | 1 | 2 |  |
| NM\_000059.4(*BRCA2*):c.2488A>G, p.Asn830Asp | [rs574039421](https://www.ncbi.nlm.nih.gov/snp/rs574039421) | Missense | Likely-benign |  |  | 1 |  |
| NM\_000059.4(*BRCA2*):c.3858\_3860del, p.Lys1286del | [rs80359406](https://www.ncbi.nlm.nih.gov/snp/rs80359406) | Synonymous | Likely-benign | 2 |  |  |  |
| NM\_000059.4(*BRCA2*):c.5986G>A, p.Ala1996Thr | [rs80358833](https://www.ncbi.nlm.nih.gov/snp/rs80358833) | Missense | Likely-benign |  |  | 1 |  |
| NM\_000059.4(*BRCA2*):c.7242A>G, p.Ser2414= | [rs1799955](https://www.ncbi.nlm.nih.gov/snp/rs1799955) | Synonymous | Benign |  | 1 | 3 |  |
| NM\_000059.4(*BRCA2*):c.7976+12G>A, g.32936842>A | [rs81002827](https://www.ncbi.nlm.nih.gov/snp/rs81002827) | Intron variant | Likely-benign |  |  | 1 |  |
| NM\_000059.4(*BRCA2*):c.8092G>A, p.Ala2698Thr | [rs80359052](https://www.ncbi.nlm.nih.gov/snp/rs80359052) | Missense | Likely-benign |  |  | 1 |  |
| NM\_000059.4(*BRCA2*):c.8487+19A>G | [rs11571743](https://www.ncbi.nlm.nih.gov/snp/rs11571743) | Intron variant | Benign |  | 1 | 1 |  |
| NM\_000059.4(*BRCA2*):c.8755-66T>C | [rs4942486](https://www.ncbi.nlm.nih.gov/snp/rs4942486) | Intron variant | Benign |  | 2 | 4 |  |
| NM\_000059.4(*BRCA2*):c.9875C>T, p.Pro3292Leu | [rs56121817](https://www.ncbi.nlm.nih.gov/snp/rs56121817) | Missense | Benign | 6 |  |  | 1 |
| NM\_000059.4(*BRCA2*):c.9976A>T, p.Lys3326Ter | [rs11571833](https://www.ncbi.nlm.nih.gov/snp/rs11571833) | Nonsense | Benign |  |  | 2 |  |
| NM\_000059.4(*BRCA2*):c.10023C>T, p.Glu3344= | [rs113507014](https://www.ncbi.nlm.nih.gov/snp/rs113507014) | Synonymous | Likely-benign | 1 |  | 1 | 1 |